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Ehlers-Danlos syndrome with recurrent bruising

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Keywords: bruising tendency, dystrophic scarring, oral blood blisters

Despite being a well-described 'textbook feature' of Ehlers-Danlos syndrome, there are few case reports or published series on bruising or bleeding in this condition. We report the case of a 62-year-old man with Ehlers-Danlos syndrome and recurrent, troublesome bruising. Clinical, haematological and biochemical features are described.

Case report

Our patient first developed skin problems in early childhood with frequent skin lacerations particularly over the lower legs, knees and elbows. These often required suturing and healed to leave large, unsightly splayed scars. Once healed, these scarred skin areas were a frequent site for haematoma formation which occurred following even trivial trauma and often took weeks to disperse. At the age of 10 years he was admitted to hospital for excision of the scarred skin overlying the knees. The wounds failed to heal satisfactorily and the scars remain widely splayed.

At 17 years he entered the army but was soon discharged because his delicate skin could not withstand the stresses applied to it during training. He was noted to suffer from severe bruising of knees and elbows for which no underlying cause could be identified at that time.

The diagnosis of Ehlers-Danlos syndrome was established in 1952 when he was first seen by a dermatologist. The following year he ruptured the left tendo-achilles which was repaired surgically, but with great difficulty. Bruising and poor wound healing were again prominent in 1967 when he underwent ligation and stripping of varicose veins from the right leg.

The past five years have seen a deterioration in his bruising tendency. He has been troubled by frequent painful bruises at pressure points on the feet which have necessitated the wearing of well cushioned shoes and walking at a gentle pace to minimize pressure and shearing forces. He has also been prone to uncomfortable bruising of the hands and fingers which have often taken weeks to disperse (Figure 1). He has recently avoided eating abrasive foods, as this has tended to induce oral blood blisters.

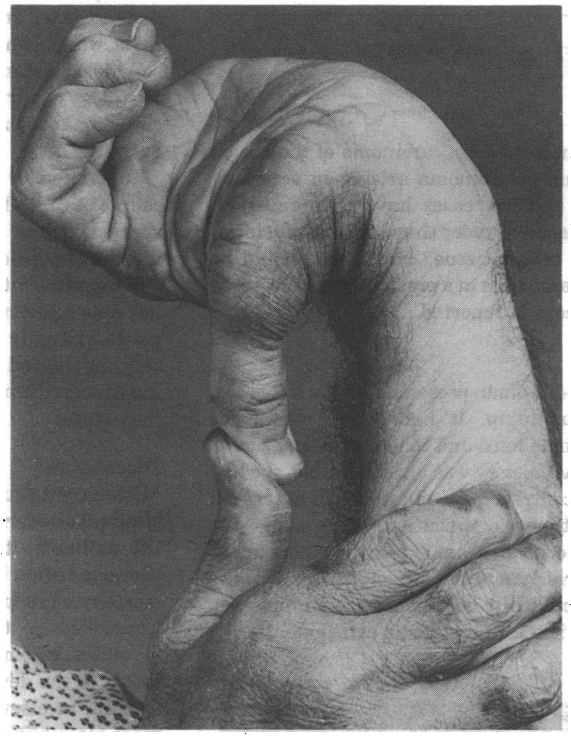

Figure 1. Hypermobile left wrist and numerous small bruises on the fingers of the right hand

Skin biopsy showed normal epidermis with some increase in dermal elastic fibres relative to a decreased amount of collagen. Full blood count was normal with a platelet count of 235×10^9/l. The prothrombin and partial thromboplatin times were both normal and no clotting factor abnormality was detected. Platelet aggregation studies showed a normal aggregation pattern to a range of concentrations of ADP, adrenaline, collagen, arachidonic acid and ristocetin. Bleeding time was normal, at 7 min, and the Hess capillary fragility test was negative. The separation of radiolabelled collagens from fibroblast culture showed no abnormalities of pro collagens nor collagen α1(I), α2(I) or α1(III) chains, therefore excluding Ehlers-Danlos syndrome types IV and VII.

Discussion

Abnormal bleeding is a well recognized feature amongst the various hereditary disorders of connective tissue. In Ehlers-Danlos syndrome (EDS) and in osteogenesis imperfecta, qualitative and quantitative abnormalities of collagen result in vascular fragility and bleeding because of deficient extravascular and perivascular support. There are, at present, 11 types recognized, of which types I, IV and X may all exhibit prominent bruising. Haematological investigation

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of type X EDS proved particularly instructive as a specific biochemical defect was identified to account for both the connective tissue and the platelet abnormalities, namely fibronectin deficiency. A number of haematological abnormalities have been reported in association with EDS including clotting factor deficiencies[1,2], capillary fragility[3], platelet ultrastructural abnormalities[4], and platelet aggregation abnormalities[5]. Abnormalities in the platelet aggregating properties of collagen in patients with EDS has also been reported[6]. There has, however, been no large series of patients, as yet, with EDS who have been investigated as regards underlying platelet or clotting abnormalities and the reports that exist do not identify any consistent defect.

In summary, we have reported a case of Ehlers-Danlos syndrome where bruising has been a prominent and troublesome feature, and in whom no underlying haematological abnormality was detected.

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Atypical X-linked variant of chronic granulomatous disease

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Keywords: chronic granulomatous disease; atypical variant

Classical X-linked recessive chronic granulomatous disease (CGD) causes recurrent severe infections in childhood. The rare atypical X-linked and autosomal recessive forms may be milder and the diagnosis overlooked. The case of a boy with an atypical variant of CGD is described.

Case report

The patient is a 7-year-old Greek-Cypriot boy of non-consanguineous parents. He presented at 3 weeks of age with bloody diarrhoea. Small pustules were present on the lower abdomen and an ulcer on the scrotum. Three months later he developed a perianal abscess which was successfully treated with antibiotics. Colonoscopy and biopsy showed non-specific colitis. Allergic colitis was suspected and he was given sulphasalazine and a proprietary milk-substitute. He subsequently underwent a milk-challenge without adverse effect. He continued to have intermittent diarrhoea and on one occasion *Clostridium difficile* was grown from his faeces. He had persistent mouth ulcers, episodes of cervical lymphadenitis, balanitis, and indolent crusted granulomatous nodules most recently on the left cheek (Figure 1). There is no history of unusual or recurrent infection in the patient's parents or his two elder sisters.

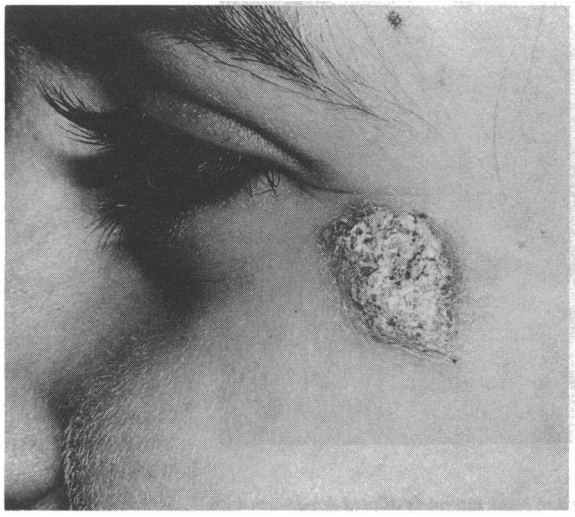

Figure 1. Crusted abscess overlying left zygoma

The infective episodes were treated with repeated courses of oral flucloxacillin and have resolved satisfactorily.

Investigations confirmed mild anaemia on occasions (minimum Hb 9.0 g/dl, Hb electrophoresis normal). His C-reactive protein (CRP) was elevated and reflected the activity of his colitis (maximum CRP 68 mg/l). Immunoglobulin levels were normal. The nitroblue tetrazolium test (NBT) was performed and was only weakly positive (NBT 0-4%), and superoxide production was detectable although greatly reduced (Table 1). The nearly normal amount of cytochrome$_{b\text{-}245}$ implies a variant form of CGD. The intermediate amount of superoxide production in his mother suggests an X-linked mode of inheritance.

Discussion

CGD is an inherited disorder of neutrophil function, in which phagocytosis is normal but in which there is a deficiency of the enzymes responsible for the burst of oxidative metabolism required to kill the ingested organisms. A defect in any part of the electron transport chain from glucose to nicotinamide adenine dinucleotide phosphate (NADPH), and then to a flavoprotein, then onto cytochrome$_{b-245}$, and finally onto oxygen will result in reduced O_2^- production and failure to kill certain ingested organisms[1].

Table 1. Superoxide production and cytochrome$_b$ concentration in the patient and his mother (courtesy of Professor A W Segal)

	Superoxide (O_2^-) production in response to PMA (nmol/min per 10^7 PMN)	*Cytochrome$_{b-245}$ (by spectroscopy) (pmol/10^7 PMN)*
Patient	4.2	74.4
Mother	46.5	129.6
Normal range	100-200	100±20

PMA, Phorbol myristate acetate; PMN, polymorphonuclear neutrophil leucocyte

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